

## Disease

# Congenital myasthenic syndrome-15<sup>1, 2</sup>

## Gene

### ALG14

|                                                 |                                                                                                                                                                                                                                                           |
|-------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                                        |
| <b>Current Treatments</b>                       | Acetylcholine-esterase inhibitors                                                                                                                                                                                                                         |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                                                            |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                       |
| <b>Non-Molecular Test</b>                       | repetitive nerve stimulation test <sup>3</sup>                                                                                                                                                                                                            |
| <b>Disease Frequency (per 100,000)</b>          | 0.92 <sup>4</sup>                                                                                                                                                                                                                                         |
| <b>Genes Included in Disease Frequency</b>      | AGRN, ALG14, SYT2, PREPL, GFPT1, SLC5A7, CHRNA1, CHRND, COLQ, DOK7, ALG2, MUSK, CHAT, SLC18A3, COL13A1, LRP4, RAPSN, DPAGT1, VAMP1, MYO9A, CHRNE, CHRNB1, SCN4A, SNAP25, SLC25A3                                                                          |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Congenital+Myasthenia&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Congenital+Myasthenia&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

## References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1168/#cms.Summary>
- <sup>2</sup> <https://www.ncbi.nlm.nih.gov/pubmed/30808424>
- <sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/30808424/>
- <sup>4</sup> <https://www.ncbi.nlm.nih.gov/pubmed/24500997>